FGF3 gene

fibroblast growth factor 3

Normal Function

The *FGF3* gene provides instructions for making a protein called fibroblast growth factor 3 (FGF3). This protein is part of a family of proteins called fibroblast growth factors that are involved in important processes such as cell division, regulation of cell growth and maturation, formation of blood vessels, wound healing, and development before birth. By attaching to another protein known as a receptor, FGF3 triggers a cascade of chemical reactions inside the cell that signal the cell to undergo certain changes, such as maturing to take on specialized functions. During development before birth, the signals triggered by the FGF3 protein stimulate cells to form the structures that make up the inner ears. The FGF3 protein is also involved in the development of many other organs and structures, including the outer ears and teeth.

Health Conditions Related to Genetic Changes

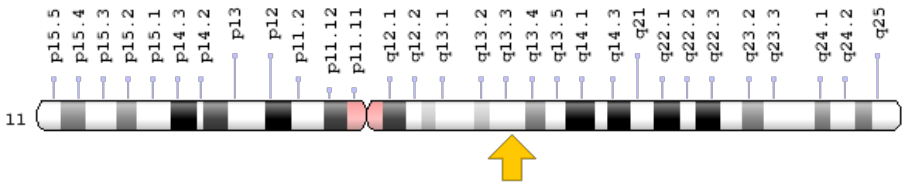
congenital deafness with labyrinthine aplasia, microtia, and microdontia

Mutations in the *FGF3* gene cause a condition known as congenital deafness with labyrinthine aplasia, microtia, and microdontia (also known as LAMM syndrome). The features of this condition include deafness caused by a lack of inner ear structures (labyrinthine aplasia), small outer ears (microtia), and small teeth (microdontia). The gene mutations involved in LAMM syndrome change single protein building blocks (amino acids) in the FGF3 protein or lead to an abnormally short protein. The altered protein likely has reduced or absent function, making it unable to stimulate signaling within cells. The loss of FGF3 function impairs development of the ears and teeth, which leads to the characteristic features of LAMM syndrome.

Chromosomal Location

Cytogenetic Location: 11q13.3, which is the long (q) arm of chromosome 11 at position 13.3

Molecular Location: base pairs 69,809,968 to 69,819,424 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FGF-3
- FGF3_HUMAN
- HBGF-3
- heparin-binding growth factor 3
- INT-2 proto-oncogene protein
- INT2
- murine mammary tumor virus integration site 2, mouse
- oncogene INT2
- proto-oncogene Int-2
- V-INT2 murine mammary tumor virus integration site oncogene homolog

Additional Information & Resources

Educational Resources

- FGF Signalling in Vertebrate Development (2010): Inner Ear
  https://www.ncbi.nlm.nih.gov/books/NBK53153/#s13.2
- Madame Curie Bioscience Database (2000): Fibroblast Growth Factors
  https://www.ncbi.nlm.nih.gov/books/NBK6330/

GeneReviews

- Congenital Deafness with Labyrinthine Aplasia, Microtia, and Microdontia
  https://www.ncbi.nlm.nih.gov/books/NBK100664

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28FGF3%5BTIAB%5D%29+OR+%28fibroblast+growth+factor+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- FIBROBLAST GROWTH FACTOR 3
  http://omim.org/entry/164950

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_FGF3.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FGF3%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3681
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2248
- UniProt
  http://www.uniprot.org/uniprot/P11487

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11066089

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